



FREM2 gene

FRAS1 related extracellular matrix protein 2

Normal Function

The *FREM2* gene provides instructions for making a protein that is part of a group of proteins called the FRAS/FREM complex; in addition to being part of the complex, FREM2 regulates the complex's formation. The FRAS/FREM complex is found in basement membranes, which are thin, sheet-like structures that separate and support cells in many tissues. The complex is particularly important during development before birth. One of its roles is to anchor the top layer of skin by connecting the basement membrane of the top layer to the layer of skin below. The FRAS/FREM complex is also involved in the proper development of certain other organs and tissues, including the kidneys, although the mechanism is unclear.

Health Conditions Related to Genetic Changes

Fraser syndrome

At least two mutations in the *FREM2* gene have been found to cause Fraser syndrome; these mutations are involved in a small percentage of cases of this condition. Fraser syndrome affects development before birth and is characterized by eyes that are completely covered by skin (cryptophthalmos), fusion of the skin between the fingers and toes (cutaneous syndactyly), and abnormalities of the kidneys and other organs and tissues.

FREM2 gene mutations involved in Fraser syndrome lead to production of an abnormal FREM2 protein that likely does not function properly. As a result, the FRAS/FREM complex cannot form. Lack of the FRAS/FREM complex in the basement membrane of skin leads to detachment of the top layer of skin, causing blisters to form during development. These blisters likely prevent the proper formation of certain structures before birth, leading to cryptophthalmos and cutaneous syndactyly. It is unknown how lack of the FRAS/FREM complex leads to kidney abnormalities and other problems in Fraser syndrome.

other disorders

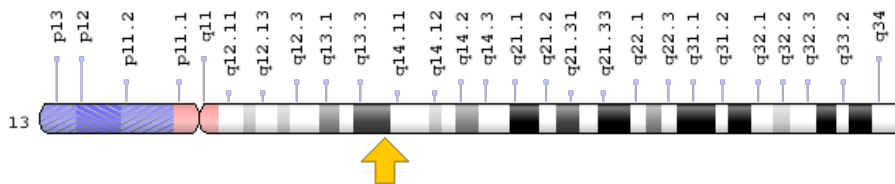
Mutations in the *FREM2* gene have also been found in people with abnormalities of the kidneys and urinary tract but no other signs and symptoms of Fraser syndrome (described above). Such abnormalities are grouped together as congenital anomalies of the kidney and urinary tract (CAKUT). The *FREM2* gene mutations involved in CAKUT typically change single protein building blocks (amino acids) in the FREM2

protein. Researchers speculate that the effects of these mutations are milder than those of mutations that cause Fraser syndrome; some FREM2 protein function may still remain. How these gene mutations affect the FRAS/FREM complex or lead to renal agenesis and other CAKUT is unknown.

Chromosomal Location

Cytogenetic Location: 13q13.3, which is the long (q) arm of chromosome 13 at position 13.3

Molecular Location: base pairs 38,687,036 to 38,887,131 on chromosome 13 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- DKFZp686J0811
- ECM3 homolog
- FRAS1-related extracellular matrix protein 2

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28FREM2%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5BIa%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- CONGENITAL ANOMALIES OF KIDNEY AND URINARY TRACT 1, SUSCEPTIBILITY TO
<http://omim.org/entry/610805>
- FRAS1-RELATED EXTRACELLULAR MATRIX PROTEIN 2
<http://omim.org/entry/608945>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_FREM2.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=FREM2%5Bgene%5D>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=25396
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/341640>
- UniProt
<http://www.uniprot.org/uniprot/Q5SZK8>

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<https://ghr.nlm.nih.gov/gene/FREM2>

Reviewed: June 2014

Published: March 21, 2017

Lister Hill National Center for Biomedical Communications
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National Institutes of Health
Department of Health & Human Services